Mouse Sox9 Sequence

1 AGTTTCAGTC CAGGAACTTT TCTTTGCAAG AGAGACGAGG TGCAAGTGGC

51 CCCGGTTTCG TTCTCTGTTT TCCCTCCCTC CTCCTCCGCT CCGACTCGCC

101 TTCCCCGGGT TTAGAGCCGG CAGCTGAGAC CCGCCACCCA GCGCCTCTGC

151 TAAGTGCCCG CCGCCGCAGC CCGGTGACGC GCCAACCTCC CCGGGAGCCG

201 TTCGCTCGGC GTCCGCGTCC GGGCAGCTGA GGGAAGAGGA GCCCCAGCCG

251 CCGCGGCTTC TCGCCTTTCC CGGCCACCCG CCCCCTGCCC CGGGCTCGCG

301 TATGAATCTC CTGGACCCCT TCATGAAGAT GACCGACGAG CAGGAGAAGG

351 GCCTGTCTGG CGCCCCCAGC CCCACCATGT CGGAGGACTC GGCTGGTTCG

401 CCCTGTCCCT CGGGCTCCGG CTCGGACACG GAGAACACCC GGCCCCAGGA

451 GAACACCTTC CCCAAGGGCG AGCCGGATCT GAAGAAGGAG AGCGAGGAAG

501 ATAAGTTCCC CGTGTGCATC CGCGAGGCGG TCAGCCAGGT GCTGAAGGGC

551 TACGACTGGA CGCTGGTGCC CATGCCCGTG CGCGTCAACG GCTCCAGCAA

601 GAACAAGCCA CACGTCAAGC GACCCATGAA CGCCTTCATG GTGTGGGCGC

651 AGGCTGCGCG CAGGAAGCTG GCAGACCAGT ACCCGCATCT GCACAACGCG

701 GAGCTCAGCA AGACTCTGGG CAAGCTCTGG AGGCTGCTGA ACGAGAGCGA

751 GAAGAGACCC TTCGTGGAGG AGGCGGAGCG GCTGCGCGTG CAGCACAAGA

801 AAGACCACCC CGATTACAAG TACCAGCCCC GGCGGAGGAA GTCGGTGAAG

851 AACGGACAAG CGGAGGCCGA AGAGGCCACG GAACAGACTC ACATCTCTCC

901 TAATGCTATC TTCAAGGCGC TGCAAGCCGA CTCCCCACAT TCCTCCTCCG

951 GCATGAGTGA GGTGCACTCC CCGGGCGAGC ACTCTGGGCA ATCTCAGGGT

1001 CCGCCGACCC CACCCACCAC TCCCAAAACC GACGTGCAAG CTGGCAAAGT

1051 TGATCTGAAG CGAGAGGGGC GCCCTCTGGC AGAGGGGGGC AGACAGCCCC

1101 CCATCGACTT CCGCGACGTG GACATCGGTG AACTGAGCAG CGACGTCATC

1151 TCCAACATTG AGACCTTCGA CGTCAATGAG TTTGACCAAT ACTTGCCACC

1201 CAACGGCCAC CCAGGGGTTC CGGCCACCCA CGGCCAGGTC ACCTACACTG

1251 GCAGTTACGG CATCAGCAGC ACCGCACCCA CCCCTGCGAC CGCGGGCCAC

Figure 1(a)

1301 GTGTGGATGT CGAAGCAGCA GGCGCCGCCC CCTCCTCCGC AGCAGCCTCC
1351 GCAGGCCCCG CAAGCCCCAC AGGCGCCTCC GCAGCAGCAA GCACCCCCGC
1401 AGCAGCCGCA GGCACCCCAG CAGCAGCAGG CACACACGCT CACCACGCTG
1451 AGCAGCGAGC CAGGCCAGTC CCAGCGAACG CACATCAAGA CGGAGCAGCT
1501 GAGCCCCAGC CACTACAGGG AGCAGCAGCA GCACTCCCCG CAACAGATCT
1551 CCTACAGCCC CTTCAACCTT CCTCACTACA GGCCCTCCTA CCCGCCCATC
1601 ACCCGTTCGG AATACGACTA CGCTGACCAT CAGAACTCCG GCTCCTACTA
1651 CAGTCACGCA GCCGGCCAGG GCTCAGGGCT CTACTCCACC TTCACTTACA
1701 TGAACCCCGC GCAGCGCCCC ATGTACACCC CCATCGGTGA CACCTCCGGG
1751 GTCCCTTCCA TCCCGCAGAC CCACAGCCCG CAGGACTGGG AACAACCAGT
1801 CTACACACAG GTCACCAGAC CCTGAGAAGA GAAAAGCTAT GGTGACAGAG
1851 CTGATCTTTT TTTTTTTTTT TTTTTAAAGA AGAAAAGAAA GAAACGAAAA
1901 AGAAAAAGCT GAAGGAAATC AAGAACCAAT TGAAATTCCT TTGGACACTT
1951 TTTTTTTTGT CCTTTCGTTA ATTTTTAAAA GACATGTAAA GGAAGGTAAC
2001 GATTGCTGGG CATTCCAGGA GAGAGACTTT AAGACTTTGT CTGAGCTCAT
2051 GACAACATAT TGCAAATGGC CGGGCCACTC GTGGCCAGAC GGACAGCACT
2101 CCTGGCCAGA TGGACCCACC AGTATCAGCG AGGAGGGGCT TGTCTCCTTC
2151 AGAGTTAACA TGGAGGACGA TTGGAGAATC TCCCTGCCTG TTTGGACTTT
2201 GTAATTATTT TTTAGCCGTA ATTAAAGAAA AAAAAAGTCC AAAAAAAAA

Figure 1(b)

Mouse Sox-9 amino acid sequence

```
Met Asn Leu Leu Asp Pro Phe Met Lys Met Thr Asp Glu Gln Glu Lys
1               5                   10                  15

Gly Leu Ser Gly Ala Pro Ser Pro Thr Met Ser Glu Asp Ser Ala Gly
            20                  25                  30

Ser Pro Cys Pro Ser Gly Ser Gly Ser Asp Thr Glu Asn Thr Arg Pro
        35                  40                  45

Gln Glu Asn Thr Phe Pro Lys Gly Glu Pro Asp Leu Lys Lys Glu Ser
    50                  55                  60

Glu Glu Asp Lys Phe Pro Val Cys Ile Arg Glu Ala Val Ser Gln Val
65                  70                  75                  80

Leu Lys Gly Tyr Asp Trp Thr Leu Val Pro Met Pro Val Arg Val Asn
                85                  90                  95

Gly Ser Ser Lys Asn Lys Pro His Val Lys Arg Pro Met Asn Ala Phe
            100                 105                 110

Met Val Trp Ala Gln Ala Ala Arg Arg Lys Leu Ala Asp Gln Tyr Pro
        115                 120                 125

His Leu His Asn Ala Glu Leu Ser Lys Thr Leu Gly Lys Leu Trp Arg
    130                 135                 140

Leu Leu Asn Glu Ser Glu Lys Arg Pro Phe Val Glu Glu Ala Glu Arg
145                 150                 155                 160

Leu Arg Val Gln His Lys Lys Asp His Pro Asp Tyr Lys Tyr Gln Pro
                165                 170                 175

Arg Arg Arg Lys Ser Val Lys Asn Gly Gln Ala Glu Ala Glu Glu Ala
            180                 185                 190

Thr Glu Gln Thr His Ile Ser Pro Asn Ala Ile Phe Lys Ala Leu Gln
        195                 200                 205

Ala Asp Ser Pro His Ser Ser Ser Gly Met Ser Glu Val His Ser Pro
    210                 215                 220

Gly Glu His Ser Gly Gln Ser Gln Gly Pro Pro Thr Pro Pro Thr Thr
225                 230                 235                 240

Pro Lys Thr Asp Val Gln Ala Gly Lys Val Asp Leu Lys Arg Glu Gly
                245                 250                 255

Arg Pro Leu Ala Glu Gly Gly Arg Gln Pro Pro Ile Asp Phe Arg Asp
            260                 265                 270
```

Figure 1(c)

```
Val Asp Ile Gly Glu Leu Ser Ser Asp Val Ile Ser Asn Ile Glu Thr
        275                 280                 285

Phe Asp Val Asn Glu Phe Asp Gln Tyr Leu Pro Pro Asn Gly His Pro
    290                 295                 300

Gly Val Pro Ala Thr His Gly Gln Val Thr Tyr Thr Gly Ser Tyr Gly
305                 310                 315                 320

Ile Ser Ser Thr Ala Pro Thr Pro Ala Thr Ala Gly His Val Trp Met
                325                 330                 335

Ser Lys Gln Gln Ala Pro Pro Pro Pro Pro Gln Gln Pro Pro Gln Ala
            340                 345                 350

Pro Gln Ala Pro Gln Ala Pro Pro Gln Gln Gln Ala Pro Pro Gln Gln
        355                 360                 365

Pro Gln Ala Pro Gln Gln Gln Gln Ala His Thr Leu Thr Thr Leu Ser
    370                 375                 380

Ser Glu Pro Gly Gln Ser Gln Arg Thr His Ile Lys Thr Glu Gln Leu
385                 390                 395                 400

Ser Pro Ser His Tyr Arg Glu Gln Gln Gln His Ser Pro Gln Gln Ile
                405                 410                 415

Ser Tyr Ser Pro Phe Asn Leu Pro His Tyr Arg Pro Ser Tyr Pro Pro
            420                 425                 430

Ile Thr Arg Ser Glu Tyr Asp Tyr Ala Asp His Gln Asn Ser Gly Ser
        435                 440                 445

Tyr Tyr Ser His Ala Ala Gly Gln Gly Ser Gly Leu Tyr Ser Thr Phe
    450                 455                 460

Thr Tyr Met Asn Pro Ala Gln Arg Pro Met Tyr Thr Pro Ile Gly Asp
465                 470                 475                 480

Thr Ser Gly Val Pro Ser Ile Pro Gln Thr His Ser Pro Gln Asp Trp
                485                 490                 495

Glu Gln Pro Val Tyr Thr Gln Val Thr Arg Pro
            500                 505
```

Figure 1(d)

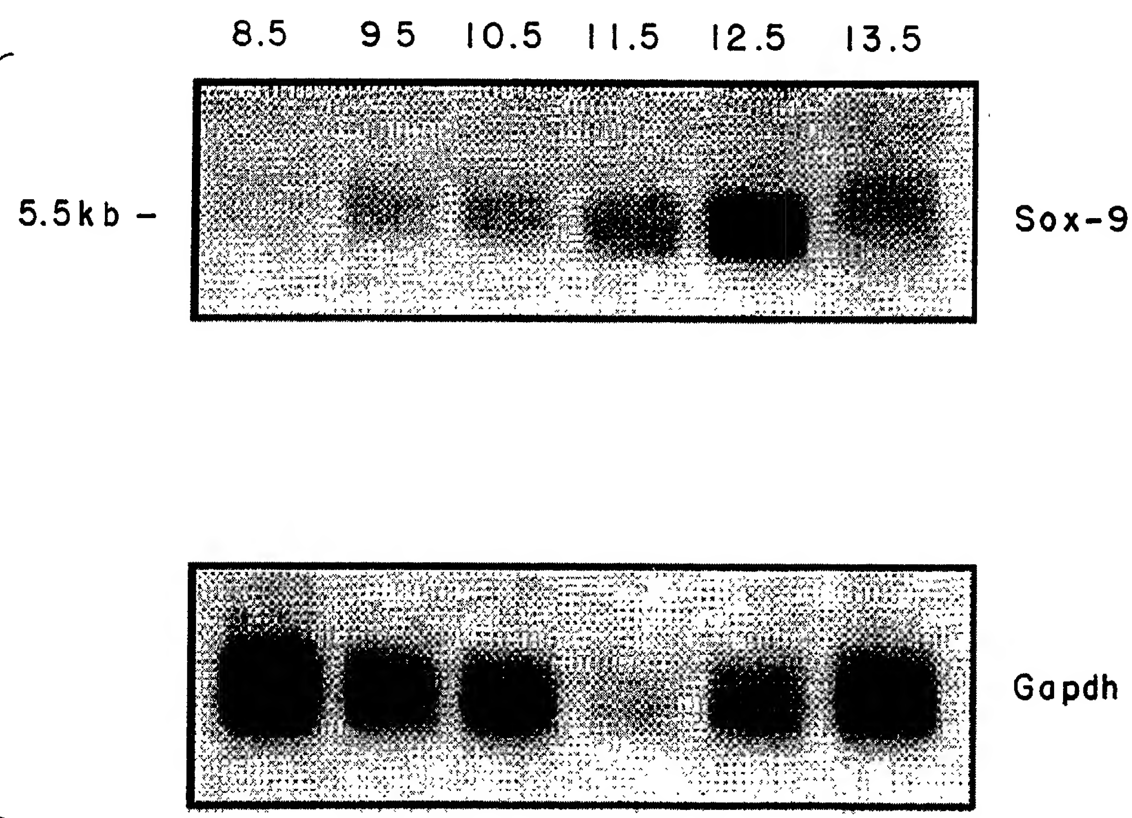


FIG.2

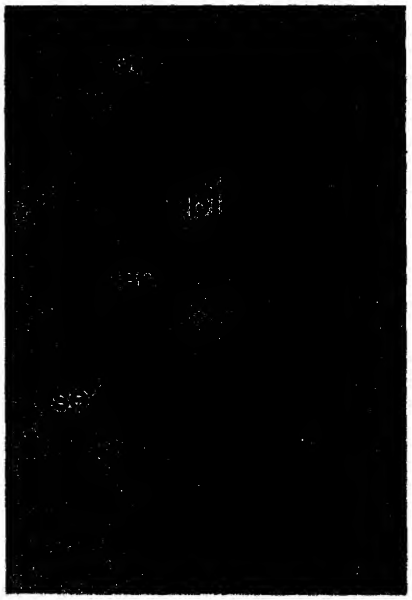


FIG.3a

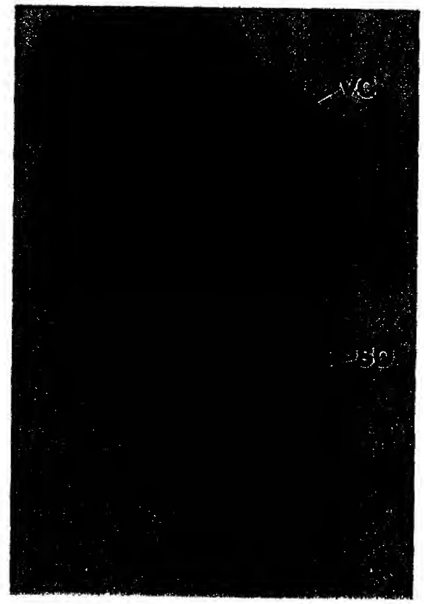


FIG.3b

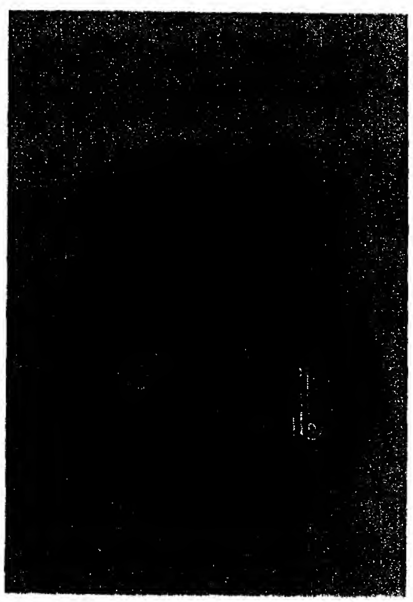


FIG.3c

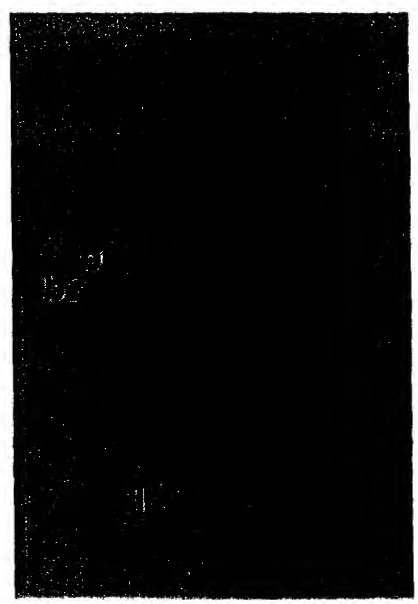


FIG.3d

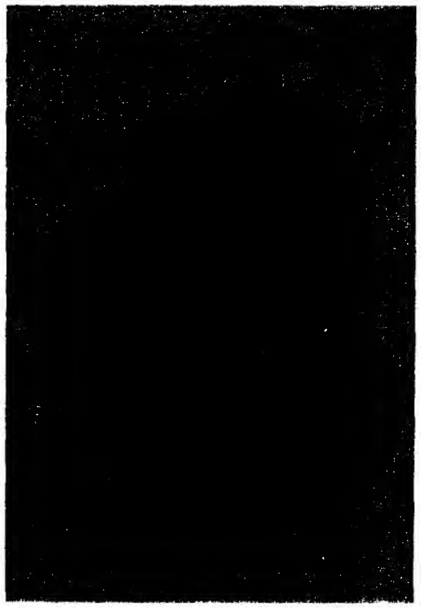


FIG.3e

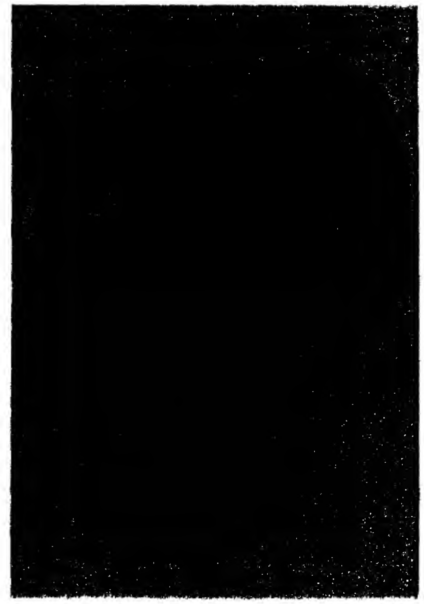


FIG.3f

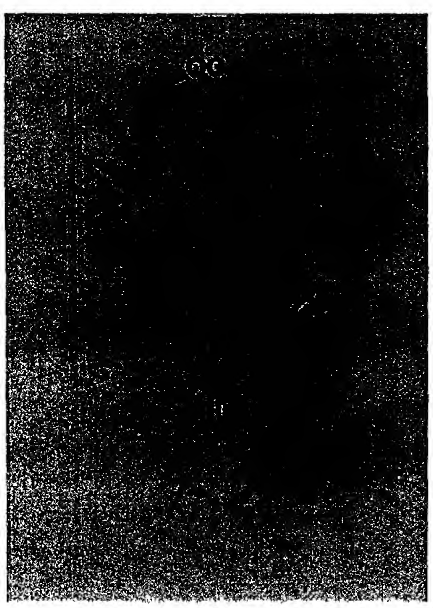


FIG.3g

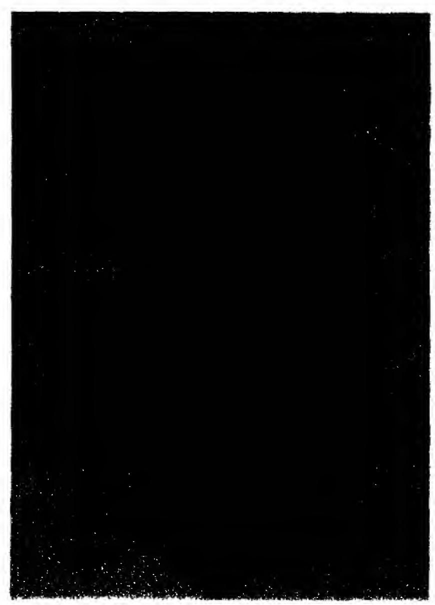


FIG.3h

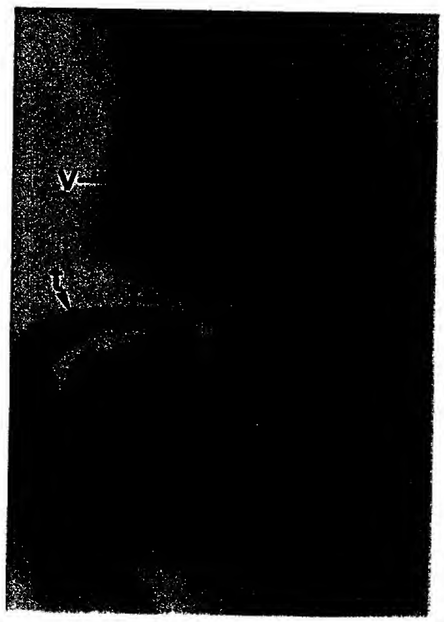


FIG.3i

FIG.4

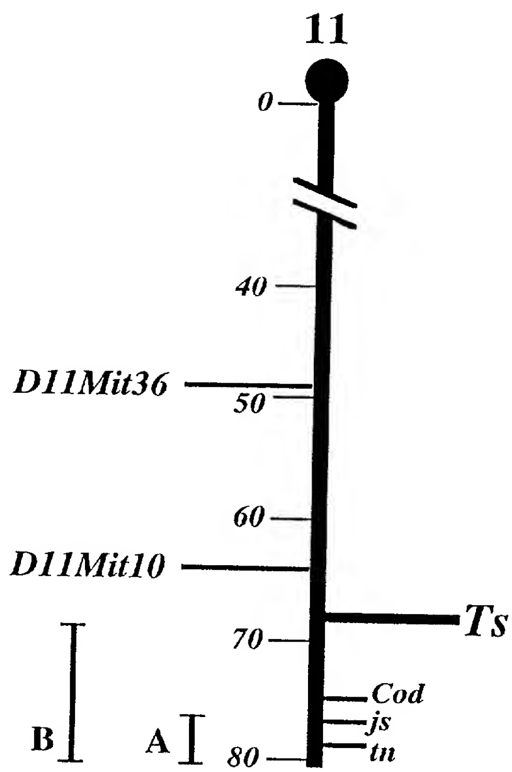


FIG.5

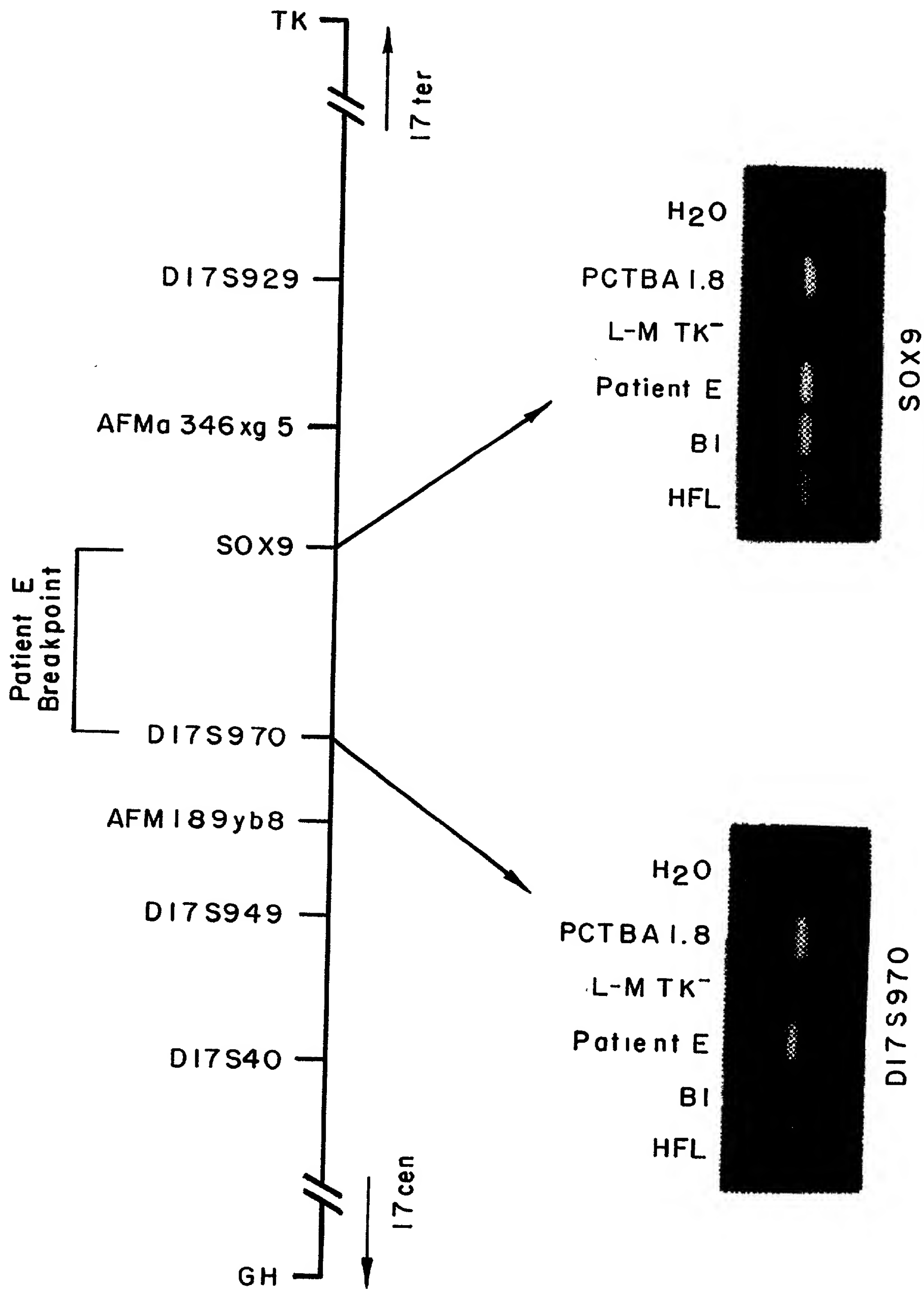
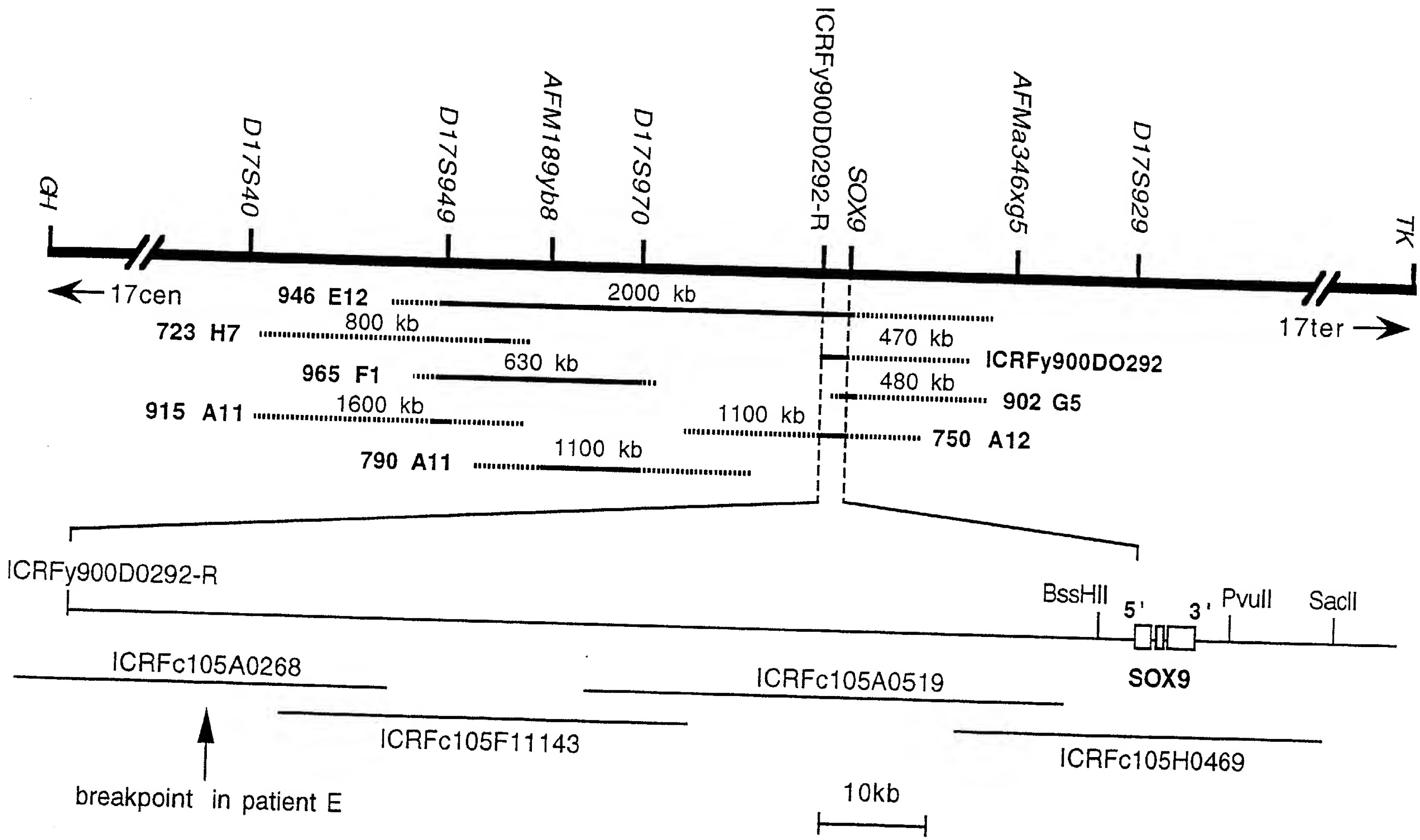

TK
17 ter
D17S929
AFMa 346xg 5
SOX9
Patient E Breakpoint
D17S970
AFM189yb8
D17S949
D17S40
17cen
GH
H_2O
PCTBA 1.8
L-M TK⁻
Patient E
B1
HFL
SOX9
H_2O
PCTBA 1.8
L-M TK⁻
Patient E
B1
HFL
D17S970

FIG.6



GH
D17S40
D17S949
AFM189yb8
D17S970
ICRFy900D0292-R
SOX9
AFMa346xg5
D17S929
TK
17cen
17ter
946 E12
2000 kb
723 H7
800 kb
470 kb
ICRFy900DO292
965 F1
630 kb
480 kb
902 G5
915 A11
1600 kb
1100 kb
750 A12
790 A11
1100 kb
ICRFy900D0292-R
BssHII
5'
3'
PvuII
SacII
SOX9
ICRFc105A0268
ICRFc105A0519
ICRFc105F11143
ICRFc105H0469
breakpoint in patient E
10kb

FIG. 7

```
CGGAGCTCGA AACTGACTGG AAACTTCAGT GGCGCGGAGA CTCGCCAGTT TCAACCCCGG
AAACTTTTCT TTGCAGGAGG AGAAGAGAAG GGGTGCAAGC GCCCCCACTT TTGCTCTTTT
TCCTCCCCTC CTCCTCCTCT CCAATTCGCC TCCCCCCACT TGGAGCGGGC AGCTGTGAAC
TGGCCACCCC GCGCCTTCCT AAGTGCTCGC CGCGGTAGCC GGCCGACGCG CCAGCTTCCC
CGGGAGCCGC TTGCTCCGCA TCCGGGCAGC CGAGGGGAGA GGAGCCCGCG CCTCGAGTCC
CCGAGCCGCC GCGGCTTCTC GCCTTTCCCG GCCACCAGCC CCCTGCCCCG GGCCCGCGTA
TGAATCTCCT GGACCCCTTC ATGAAGATGA CCGACGAGCA GGAGAAGGGC CTGTCCGGCG
CCCCCAGCCC CACCATGTCC GAGGACTCCG CGGGCTCGCC CTGCCCGTCG GGCTCCGGCT
CGGACACCGA GAACACGCGG CCCCAGGAGA ACACGTTCCC CAAGGGCGAG CCCGATCTGA
AGAAGGAGAG CGAGGAGGAC AAGTTCCCCG TGTGCATCCG CGAGGCGGTC AGCCAGGTGC
TCAAAGGCTA CGACTGGACG CTGGTGCCCA TGCCGGTGCG CGTCAACGGC TCCAGCAAGA
ACAAGCCGCA CGTCAAGCGG CCCATGAACG CCTTCATGGT GTGGGCGCAG GCGGCGCGCA
GGAAGCTCGC GGACCAGTAC CCGCACTTGC ACAACGCCGA GCTCAGCAAG ACGCTGGGCA
AGCTCTGGAG ACTTCTGAAC GAGAGCGAGA AGCGGCCCTT CGTGGAGGAG GCGGAGCGGC
TGCGCGTGCA GCACAAGAAG GACCACCCGG ATTACAAGTA CCAGCCGCGG CGGAGGAAGT
CGGTGAAGAA CGGGCAGGCG GAGGCAGAGG AGGCCACGGA GCAGACGCAC ATCTCCCCCA
ACGCCATCTT CAAGGCGCTG CAGGCCGACT CGCCACACTC CTCCTCCGGC ATGAGCGAGG
TGCACTCCCC CGGCGAGCAC TCGGGGCAAT CCCAGGGCCC ACCGACCCCA CCCACCACCC
CCAAAACCGA CGTGCAGCCG GGCAAGGCTG ACCTGAAGCG AGAGGGGCGC CCCTTGCCAG
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ACGTCATCTC CAACATCGAG ACCTTCGATG TCAACGAGTT TGACCAGTAC CTGCCGCCCA
ACGGCCACCC GGGGGTGCCG GCCACGCACG GCCAGGTCAC CTACACGGGC AGCTACGGCA
TCAGCAGCAC CGCGGCCACC CCGGCGAGCG CGGGCCACGT GTGGATGTCC AAGCAGCAGG
CGCCGCCGCC ACCCCCGCAG CAGCCCCCAC AGGCCCCGCC GGCCCCGCAG GCGCCCCCGC
AGCCGCAGGC GGCGCCCCCA CAGCAGCCGG CGGCACCCCC GCAGCAGCCA CAGGCGCACA
CGCTGACCAC GCTGAGCAGC GAGCCGGGCC AGTCCCAGCG AACGCACATC AAGACGGAGC
AGCTGAGCCC CAGCCACTAC AGCGAGCAGC AGCAGCACTC GCCCCAACAG ATCGCCTACA
GCCCCTTCAA CCTCCCACAC TACAGCCCCT CCTACCCGCC CATCACCCGC TCACAGTACG
ACTACACCGA CCACCAGAAC TCCAGCTCCT ACTACAGCCA CGCGGCAGGC CAGGGCACCG
GCCTCTACTC CACCTTCACC TACATGAACC CCGCTCAGCG CCCCATGTAC ACCCCCATCG
CCGACACCTC TGGGGTCCCT TCCATCCCGC AGACCCACAG CCCCCAGCAC TGGGAACAAC
CCGTCTACAC ACAGCTCACT CGACCTTGAG GAGGCCTCCC ACGAAGGGCG ACGATGGCCG
AGATGATCCT AAAAATAACC GAAGAAAGAG AGGACCAACC AGAATTCCCT TTGGACATTT
GTGTTTTTTT GTTTTTTTAT TTTGTTTTGT TTTTTCTTCT TCTTCTTCTT CCTTAAAGAC
ATTTAAGCTA AAGGCAACTC GTACCCAAAT TTCCAAGACA CAAACATGAC CTATCCAAGC
GCATTACCCA CTTGTGGCCA ATCAGTGGCC AGGCCAACCT TGGCTAAATG GAGCAGCGAA
ATCAACGAGA AACTGGACTT TTTAAACCCT CTTCAGAGCA AGCGTGGAGG ATGATGGAGA
ATCGTGTGAT CAGTGTGCTA AATCTCTCTG CCTGTTTGGA CTTTGTAATT ATTTTTTTAG
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TGTACTGTGT ATGATTCATT ACCATTTTGA GGGGATTTAT ACATATTTTT AGATAAAATT
AAATGCTCTT ATTTTTCCAA CAGCTAAACT ACTCTTAGTT GAACAGTGTG CCCTAGCTTT
TCTTGCAACC AGAGTATTTT TGTACAGATT TGCTTTCTCT TACAAAAAGA AAAAAAAAAT
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TATATATATT TTTTAAAGAA GAGAAAAACA CCTTGAGCCT TAAAACGGTG CTGCTGGGAA
ACATTTGCAC TCTTTTAGTG CATTTCCTCC TGCCTTTGCT TGTTCACTGC AGTCTTAAGA
AAGAGGTAAA AGGCAAGCAA AGGAGATGAA ATCTGTTCTG GGAATGTTTC AGCAGCCAAT
AAGTGCCCGA GCACACTGCC CCCGGTTGCC TGCCTGGGCC CCATGTGGAA GGCAGATGCC
TGCTCGCTCT GTCACCTGTG CCTCTCAGAA CACCAGCAGT TAACCTTCAA GACATTCCAC
```

Figure 8a(1)

TTGCTAAAAT TATTTATTTT GTAAGGAGAG GTTTTAATTA AAACAAAAAA AAATTCTTTT
TTTTTTTTTT TTTTCCAATT TTACCTTCTT TAAAATAGGT TGTTGGAGCT TTCCTCAAAG
GGTATGGTCA TCTGTTGTTA AATTATGTTC TTAACTGTAA CCAGTTTTTT TTTATTTATC
TCTTTAATCT TTTTTATTAT TAAAAGCAAG TTTCTTTGTA TTCCTCACCC TAGATTTGTA
TAAATGCCTT TTTGTCCATC CCTTTTTTCT TTGTTGTTTT TGTTGAAAAC AAACTGGAAA
CTTGTTTCTT TTTTTGTATA AATGAGAGAT TGCAAATGTA GTGTATCACT GAGTCATTTG
CAGTGTTTTC TGCCACAGAC CTTTGGGCTG CCTTATATTG TGTGTGTGTG TGGGTGTGTG
TGTGTTTTGA CACAAAAACA ATGCAAGCAT GTGTCATCCA TATTTCTCTA CATCTTCTCT
TGGAGTGAGG GAGGCTACCT GGAGGGGATC AGCCCACTGA CAGACCTTAA TCTTAATTAC
TGCTGTGGCT AGAGAGTTTG AGGATTGCTT TTTAAAAAAG ACAGCAAACT TTTTTTTTTA
TTTAAAAAAA GATATATTAA CAGTTTTAGA AGTCAGTAGA ATAAAATCTT AAAGCACTCA
TAATATGGCA TCCTTCAATT TCTGTATAAA AGCAGATCTT TTTAAAAAAG ATACTTCTGT
AACTTAAGAA ACCTGGCATT TAAATCATAT TTTGTCTTTA GGTAAAAGCT TTGGTTTGTG
TTCGTGTTTT GTTTGTTTCA CTTGTTTCCC TCCCAGCCCC AAACCTTTTG TTCTCTCCGT
GAAACTTACC TTTCCCTTTT TCTTTCTCTT TTTTTTTTTG TATATTATTG TTTACAATAA
ATATACATTG CATTAAAAAG AAA

Figure 8a(2)

Met Asn Leu Leu Asp Pro Phe Met Lys Met Thr Asp Glu Gln Glu Lys
1 5 10 15
Gly Leu Ser Gly Ala Pro Ser Pro Thr Met Ser Glu Asp Ser Ala Gly
20 25 30
Ser Pro Cys Pro Ser Gly Ser Gly Ser Asp Thr Glu Asn Thr Arg Pro
35 40 45
Gln Glu Asn Thr Phe Pro Lys Gly Glu Pro Asp Leu Lys Lys Glu Ser
50 55 60
Glu Glu Asp Lys Phe Pro Val Cys Ile Arg Glu Ala Val Ser Gln Val
65 70 75 80
Leu Lys Gly Tyr Asp Trp Thr Leu Val Pro Met Pro Val Arg Val Asn
85 90 95
Gly Ser Ser Lys Asn Lys Pro His Val Lys Arg Pro Met Asn Ala Phe
100 105 110
Met Val Trp Ala Gln Ala Ala Arg Arg Lys Leu Ala Asp Gln Tyr Pro
115 120 125
His Leu His Asn Ala Glu Leu Ser Lys Thr Leu Gly Lys Leu Trp Arg
130 135 140
Leu Leu Asn Glu Ser Glu Lys Arg Pro Phe Val Glu Glu Ala Glu Arg
145 150 155 160
Leu Arg Val Gln His Lys Lys Asp His Pro Asp Tyr Lys Tyr Gln Pro
165 170 175
Arg Arg Arg Lys Ser Val Lys Asn Gly Gln Ala Glu Ala Glu Glu Ala
180 185 190
Thr Glu Gln Thr His Ile Ser Pro Asn Ala Ile Phe Lys Ala Leu Gln
195 200 205
Ala Asp Ser Pro His Ser Ser Ser Gly Met Ser Glu Val His Ser Pro
210 215 220
Gly Glu His Ser Gly Gln Ser Gln Gly Pro Pro Thr Pro Pro Thr Thr
225 230 235 240
Pro Lys Thr Asp Val Gln Pro Gly Lys Ala Asp Leu Lys Arg Glu Gly
245 250 255
Arg Pro Leu Pro Glu Gly Gly Arg Gln Pro Pro Ile Asp Phe Arg Asp
260 265 270
Val Asp Ile Gly Glu Leu Ser Ser Asp Val Ile Ser Asn Ile Glu Thr
275 280 285
Phe Asp Val Asn Glu Phe Asp Gln Tyr Leu Pro Pro Asn Gly His Pro
290 295 300
Gly Val Pro Ala Thr His Gly Gln Val Thr Tyr Thr Gly Ser Tyr Gly
305 310 315 320
Ile Ser Ser Thr Ala Ala Thr Pro Ala Ser Ala Gly His Val Trp Met
325 330 335
Ser Lys Gln Gln Ala Pro Pro Pro Pro Pro Gln Gln Pro Pro Gln Ala
340 345 350
Pro Pro Ala Pro Gln Ala Pro Pro Gln Pro Gln Ala Ala Pro Pro Gln
355 360 365
Gln Pro Ala Ala Pro Pro Gln Gln Pro Gln Ala His Thr Leu Thr Thr
370 375 380

Figure 8a(3)

```
Leu Ser Ser Glu Pro Gly Gln Ser Gln Arg Thr His Ile Lys Thr Glu
385                 390                 395                 400
Gln Leu Ser Pro Ser His Tyr Ser Glu Gln Gln Gln His Ser Pro Gln
                405                 410                 415
Gln Ile Ala Tyr Ser Pro Phe Asn Leu Pro His Tyr Ser Pro Ser Tyr
            420                 425                 430
Pro Pro Ile Thr Arg Ser Gln Tyr Asp Tyr Thr Asp His Gln Asn Ser
        435                 440                 445
Ser Ser Tyr Tyr Ser His Ala Ala Gly Gln Gly Thr Gly Leu Tyr Ser
    450                 455                 460
Thr Phe Thr Tyr Met Asn Pro Ala Gln Arg Pro Met Tyr Thr Pro Ile
465                 470                 475                 480
Ala Asp Thr Ser Gly Val Pro Ser Ile Pro Gln Thr His Ser Pro Gln
                485                 490                 495
His Trp Glu Gln Pro Val Tyr Thr Gln Leu Thr Arg Pro
            500                 505
```

Figure 8a(4)

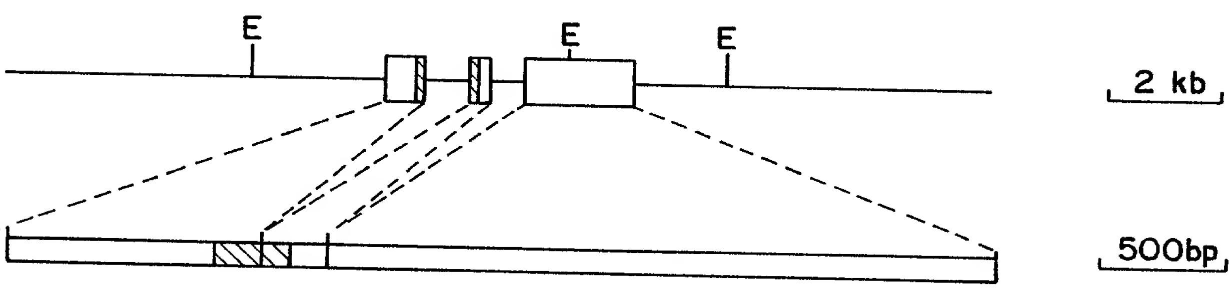

FIG.8b

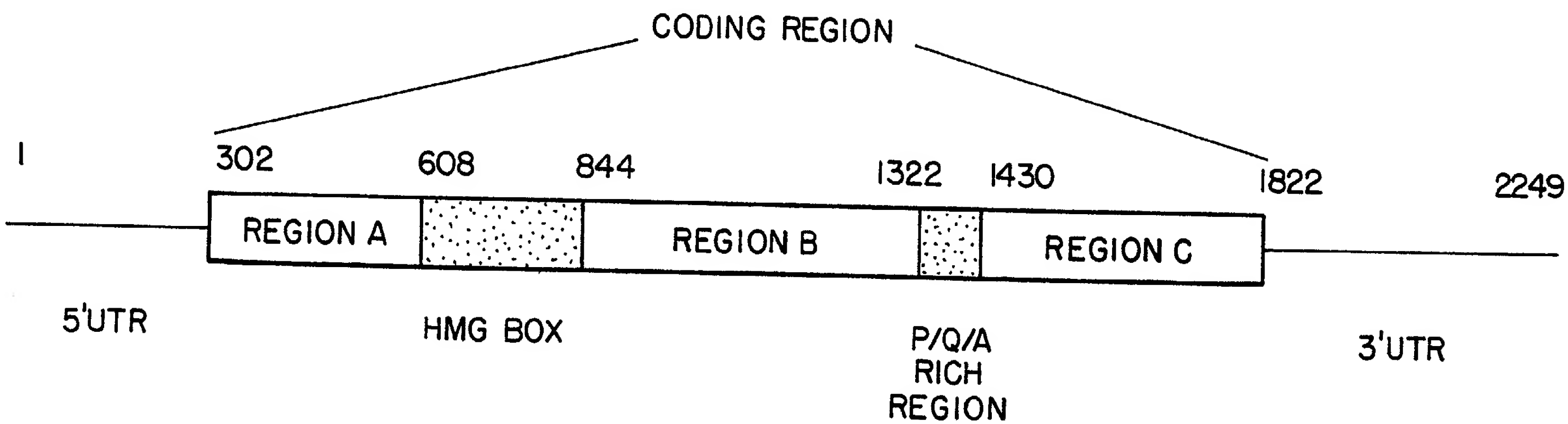
CODING REGION
1
302
608
844
1322
1430
1822
2249
REGION A
REGION B
REGION C
5'UTR
HMG BOX
P/Q/A
RICH
REGION
3'UTR

FIG.9

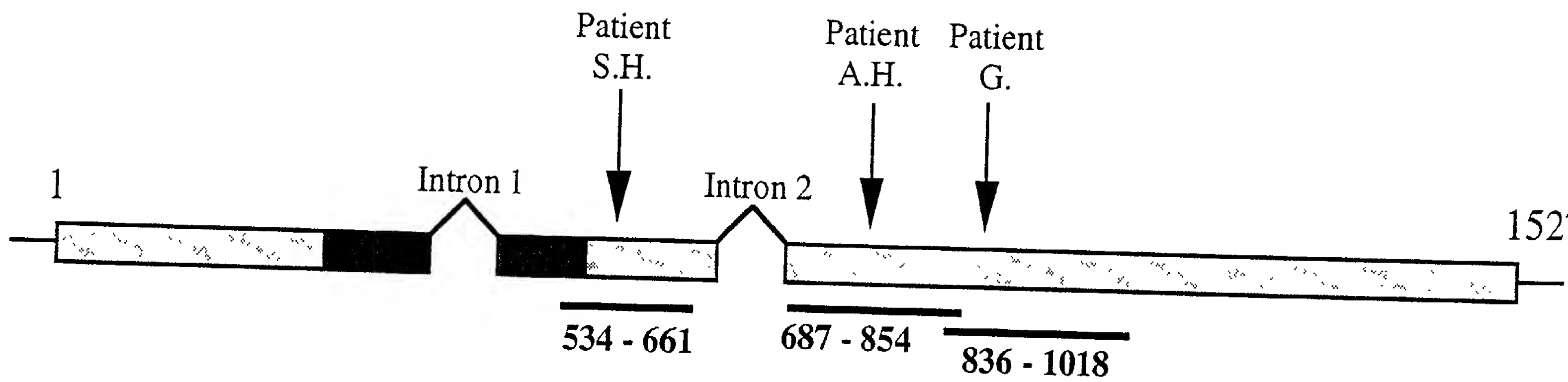
Patient
S.H.
Patient
A.H.
Patient
G.
1
Intron 1
Intron 2
1527
534 - 661
687 - 854
836 - 1018

FIG.10a

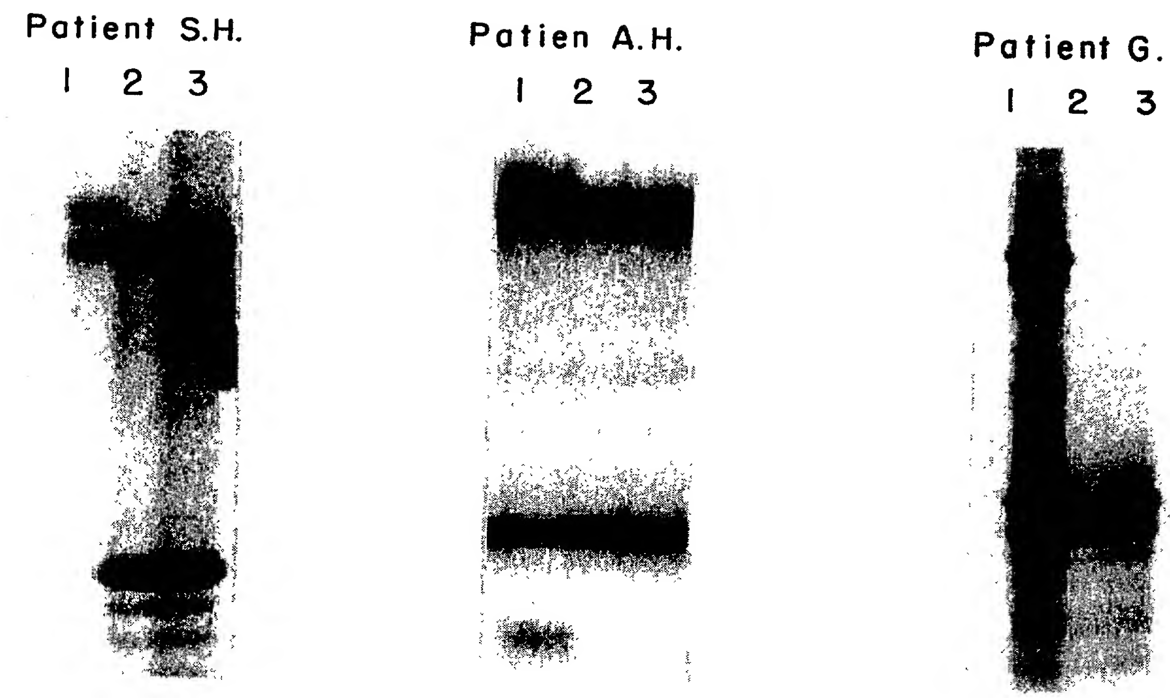

FIG.10b

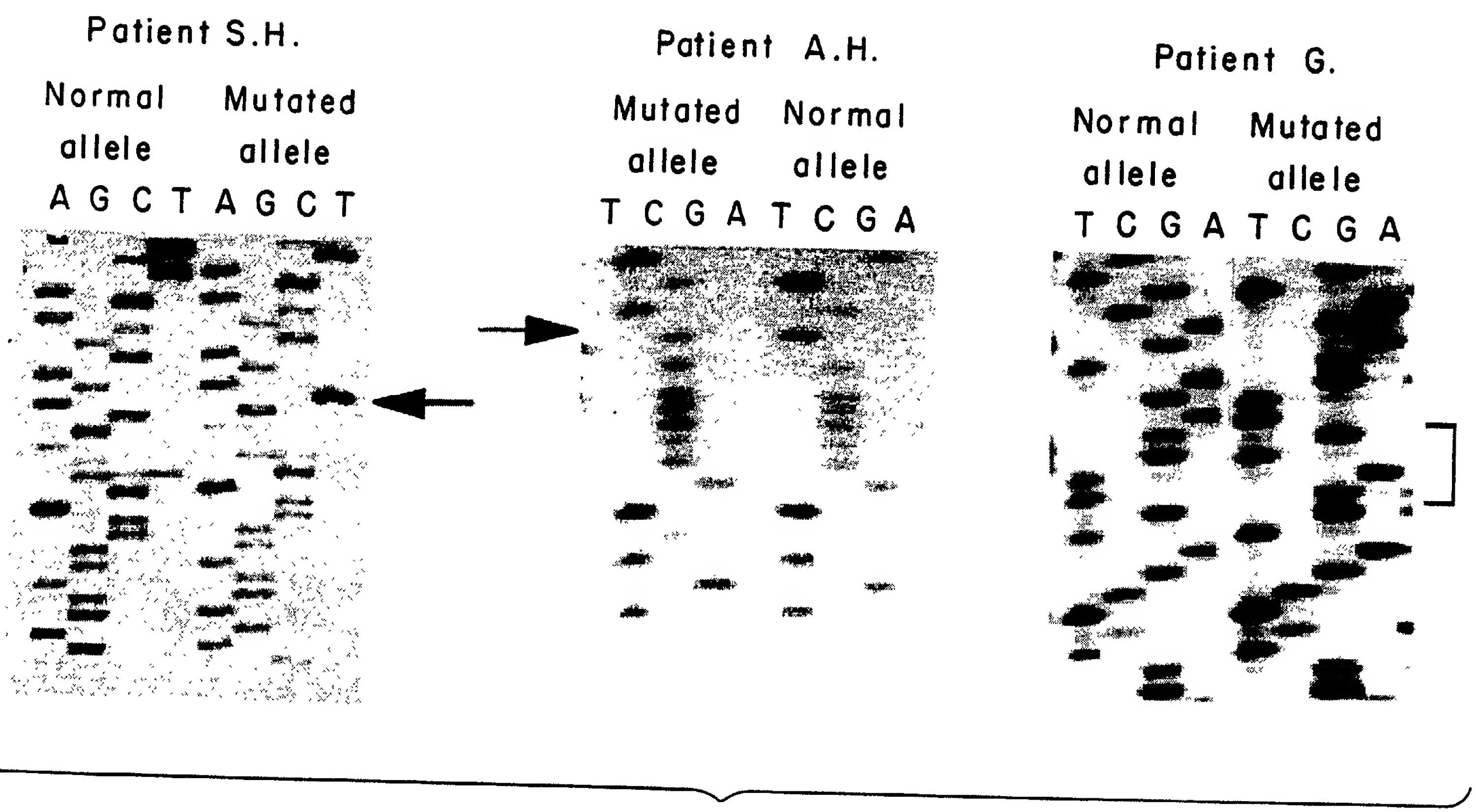
Patient S.H.
Normal allele
Mutated allele
A G C T A G C T
Patient A.H.
Mutated allele
Normal allele
T C G A T C G A
Patient G.
Normal allele
Mutated allele
T C G A T C G A

FIG.10c